FREQUENTLY ASKED QUESTIONS

MJD FOUNDATION

ABOUT THE DISEASE?

What is Machado-Joseph Disease?

Machado-Joseph Disease (MJD) is a hereditary neuro-degenerative condition. MJD occurs because of a fault on chromosome 14q that results in the production of too much of an abnormal protein known as 'Ataxin 3'. This protein causes nerve cells to die prematurely in a part of the brain called the cerebellum, along with the brainstem. The damage to the cerebellum and brainstem initially causes muscular weakness and progresses over time to a total lack of voluntary control and very significant permanent physical disability.

What other disease is MJD similar to?

MJD is in a 'family' of neuro-degenerative diseases that includes Huntington's Disease and is one of the spino-cerebellar ataxias (SCAs). MJD is also known as SCA3. Many MJD symptoms are very similar to Huntington's. The most significant difference is that there is limited cognitive deterioration; although recent literature suggests that there may be some high level executive processing deficits, in practical terms people with MJD remain intellectually and emotionally intact throughout the progression of the disease (see below).

Are people with MJD intellectually affected?

No, people with MJD generally retain their cognitive capacity and intellectual function for the entire progression of the disease. This is one of the most misunderstood parts of the disease, as people lose their ability to speak, and this along with symptoms such as spasticity and tremor often gives the impression of an intellectual impairment. It is important however to be aware of very high rates of depressive illness among people who have MJD and this may impact decision making capacity and engagement.

Is there a cure for MJD?

No, currently there is no known cure for MJD. Progression to dependence occurs over 5 to 10 years and most people are wheelchair bound and fully dependent for activities of daily living within 10 years of the first symptoms emerging.

What are the initial/early symptoms of MJD?

This can differ from person to person, however the first symptoms of MJD are usually related to ataxia which results in feeling unbalanced (feeling like falling over) – especially at night to start with; problems with vision (double vision called diplopia or shaky eyes called nystagmus); and problems with strength in arms or legs.

Does the progression of MJD follow a predictable pattern?

The exact progression is different from person to person, as is the speed of the progression. However, the pattern seen is a gradual degeneration in:

- ability to walk without falling over;
- verbal communication ability;
- swallowing ability;
- vision (blurred and shaky vision);
- speed of reflexes (or reaction time);
- muscle strength; and
- urinary continence.

What does a person with MJD usual die from?

This differs greatly from person to person. Some causes of death are: aspiration pneumonia, bowel obstruction, infection or malnutrition.

How long does it take from first symptoms to needing full time care?

This differs greatly from person to person and it also depends on how long the defect (mutation) in the affected gene is. The younger generations of a family who have MJD may start to experience symptoms earlier than their parents

FREQUENTLY ASKED QUESTIONS

MJD FOUNDATION

and become Therefore, it could be anywhere from 5 years to 20 years.

Are there any treatments for MJD?

There are some treatments for symptoms of MJD such as muscle cramping, poor sleep, balance, pain etc. The MJD Foundation is working on a set of medical protocols to bring together international best practice on treating symptoms of MJD. These may be found on the "Resources" tab of our website.

What kinds of therapeutic support are helpful for people with MJD?

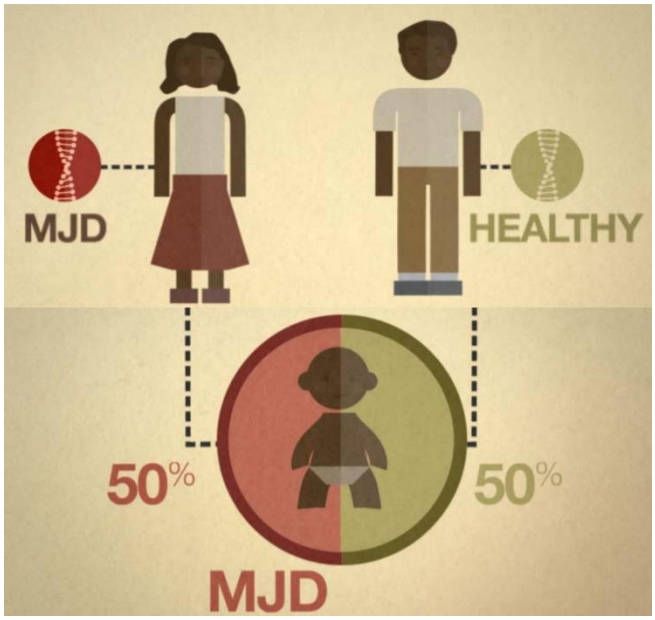
Any exercises to keep muscles strong, and in particular keep good core strength may help to slow the progression of MJD. The MJD Foundation recommends regular physiotherapy and hydrotherapy, and encourages all sorts of exercise from bike riding to fishing. Massage and chelated magnesium also appears to assist with muscle cramping, although the scientific evidence here is inconclusive. There is research currently being conducted into the amount of exercise that is best for people who have MJD.

How should I refer to a person who has MJD?

We don't like to use the word "sufferer" as this labels a person as "suffering" for their entire life's journey with MJD. Instead we prefer to say "person with MJD" or "person living with MJD".

ABOUT INHERITANCE OF MJD

How does a person get MJD? You must have a parent (either Mum or Dad) who has the disease. MJD is an inherited, autosomal dominant disorder, meaning that each child of a person who carries the defective gene has a 50% chance of developing the disease. In addition, the mutation is typically expanded (worse) when it is passed to the next generation (known as an 'anticipation effect'). This means that symptoms of the disease can appear (but not always) around 8 years to 10 years earlier and progress more rapidly



If people have the affected gene, do they always develop symptoms or do they sometimes carry the gene without showing symptoms themselves? A normal MJD gene should have 12-44 'repeat sequences' if a person has an MJD gene expansion of over 61 repeats, they will definitely have the disease and will develop symptoms at some stage during normal life expectancy. This is called "Autosomal Dominant".

Can a sibling that does not have MJD pass it onto their own children?

No. There is no "carrier" concept with MJD.

Why do the children of people with the disease often experience symptoms at a younger age than their parent?

Although this does not always happen, it is common. When the disease is passed from parent to child, the mutation on the gene is frequently expanded (or worse). This is called the "Anticipation Effect". In basic terms, the larger the mutation, the earlier the age of onset.

FREQUENTLY ASKED QUESTIONS

MJD FOUNDATION

So, if the gene is passed on earlier and worse to the child, why doesn't MJD eventually 'die out' in families, as the person with the disease gets very sick before child bearing age?

In some cases where a teenage child develops MJD, it is possible that the disease will 'die out' in that line of the family; however, the anticipation affect is not always predictable, and where one sibling may develop the disease in teenage years, another affected sibling may not exhibit symptoms until later in life. A person with MJD has CAG repeats between 51 and 86. There is no "carrier" concept with MJD as there is with some other types of genetic diseases, but people who have the gene expansion of between 44 and 61 repeats may appear normal (have no symptoms) but pass on a further (longer) expansion to their children. This is why we think the disease has been in the human population for thousands of years.

Does MJD affect men or women more?

There is no conclusive scientific evidence to suggest that either men or women are more disposed to inheriting the MJD gene.

Why does one sibling get the disease and another sibling not get it?

Because of the way genetics work; it is left to chance (luck/bad luck) as to which of your Mum's or Dad's genes you inherit. It is a cruel fact of the disease that the unaffected sibling will often feel enormous guilt at not having inherited the disease, whilst their sibling has.

What is a CAG length or Allelle length?

Everyone inherits their chromosomes in pairs (there are 23 in total); one chromosome from their Mum and one from their Dad. This makes up one small part of our DNA and determines our characteristics, just like our hair or skin colour. A gene is made up of combinations of 4 proteins called A, G, T and C. There is a very specific number of combinations of the 4 proteins needed for each gene. People with MJD have a problem with too many CAG, combinations, this 'repeats' too many times on chromosome (Allele) 14q. A normal number of CAG repeats is 43. A person with MJD has CAG repeats between 51 and 86.

CARING FOR SOMEONE WITH MJD

What are some of the things that challenge family carers of people with MJD?

There are many complex challenges for family carers. People with MJD eventually require 24 hour care because they require assistance for all activities of daily living and they also have high levels of sleep disturbance. The biggest complaint from carers is that they are exhausted through lack of sleep themselves and that they have difficulty understanding what their loved one is trying to communicate. Adding to the difficulties for family carers is that in many cases the carer may be a child of the person they care for and they may also have early signs of MJD, (like balance or vision problems) or be caring for more than one generation of people with MJD at the same time. This puts an enormous physical, emotional and economic burden onto carers.

In a residential care setting, what are some of the issues that challenge people with MJD and their carers?

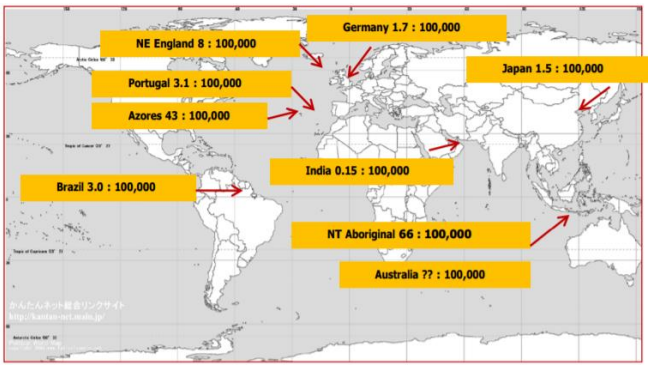
People with MJD can still think, and like to be in control of their lives. Often their bodies can still perform some basic movements, but much more slowly and more clumsily than others. This is especially evident when people try to talk or communicate with an assistive device like an iPad. Everything takes a very long time and some things are just impossible independently. This is very frustrating. It is important to understand that allowing enough time is very important. In residential care environments people with MJD communicate their extreme frustration and shame at often

being treated as though they are intellectually disabled (when in fact their intellect is intact). Their problems with communication and swallowing require carers to take time and be patient, and there are high levels of frustration experienced when this does not occur.

WHERE DOES MJD OCCUR?

Does MJD only affect Aboriginal Australians?

No, MJD is found all over the world and affects people of different races and ethnicities. Where else is it found internationally? Almost everywhere, with large incidences in Portugal, Brazil, China, India, Japan, and Australia.



How did the disease come into the Aboriginal population of the NT?

Spread of MJD to Arnhem Land had previously (before Feb 2012) been attributed to the 16th Century trading and exploration activities of Portuguese Sailors. Entry into the Australian population was thought to have been through trading relationships between the Aboriginal people of Arnhem Land and the Macassan people of Indonesia, who in turn traded with the Portuguese. However in February 2012, research was published which effectively rules out a Portuguese link, and instead points to a direct Asian link based on an international DNA 'Haplotyping' study. This means that the type of MJD experienced in Northern Australia is most like the Asian variant of the disease and this is known to favour an aggressive anticipation effect. We know therefore that more people will develop the disease earlier than their parents in this population.

Where are Aboriginal Australians living with MJD?

Groote Eylandt • Angurugu • Umbakumba

Bickerton Island • Milyakburra

Elcho Island • Galiwin'ku

Mainland – Top End • Yirrkala • Birany Birany • Numbulwar • Ngukurr • Oenpelli • Darwin

Mainland – Central Australia • Papunya • Hermannsburg (Ntaria) • Santa Teresa

Mainland – Queensland • Cairns • Kuranda • Atherton

FREQUENTLY ASKED QUESTIONS

MJD FOUNDATION

What are the current numbers of Aboriginal Australians with MJD? (As per records that the MJD Foundation has established as at March 2017)

Location	Symptomatic or +'ve test	Monitored ^	At risk *
Groote Eylandt	12	13	186
Yirrkala	3	14	112
Elcho Island	11	12	121
Ngukurr	5	10	95
Central Australia	5	3	97
Oenpelli	3	1	8
Nth Qld	5	1	23
S. Australia	1	0	0
	46	56	646

^ Monitored – are people who have had symptoms reported and are being monitored by the MJDF – we calculate that 80% of these people have MJD * At risk – are people alive today who have at least a 50% chance of having MJD. Based on the above numbers and the 20% estimated error rate for those in the monitored column, there are currently 92 clients of the MJDF who have MJD.

THE MJD FOUNDATION

Who does the MJD Foundation help?

The MJD Foundation was established as a charity in 2008. The MJD Foundation works in partnership with Aboriginal[1] Australians, their families and communities living with MachadoJoseph Disease to provide comprehensive supports and engage in research to provide hope for the future.

Why does the MJD Foundation focus on Aboriginal Australians?

A few reasons:

1. Aboriginal Australians living in the Northern Territory have a prevalence of 100x greater than the international average. 2. Many Aboriginal Australians with MJD also live in remote communities where there are significant gaps in service compared with those available in a non-remote setting.
2. Our current funding comes predominantly from the Aboriginals Benefit Account (NT) and the Anindilyakwa Land Council on Groote Eylandt.

Can the MJD Foundation help me if I am not Aboriginal?

Yes, we can help in the following ways:

1. We sometimes have some small grant funding to help with equipment for non Indigenous Australians
2. We can recommend who the Australian MJD medical specialists are
3. We will share any information we can, such as Medical Protocols, research findings etc
4. We will put your GP in contact with MJD specialists to help you get the best treatment possible.

[1] The MJDF uses the term Aboriginal in preference to Aboriginal and Torres Strait Islander in recognition that Aboriginal people are the original inhabitants of Arnhem Land. Aboriginal and Torres Strait Islander is referred to in the national context. No disrespect is intended to our Torres Strait Islander families and communities. The MJDF acknowledges the inclusion of Torres Strait Islanders who may be affected by MJD.

FREQUENTLY ASKED QUESTIONS

MJD FOUNDATION

When was the MJD Foundation established?
The MJD Foundation was established as a charity under the NT Associations Act in May 2008. In 2012 the MJD Foundation transitioned to be a public company limited by guarantee under the Australia Corporations Act. The MJD Foundation is now governed by the Charities Act under the ACNC.

Where does the MJD Foundation get its funding?
We have received most of our funding from the Aboriginals Benefit Account (which is a mining royalty matched fund that the federal government administers) and the Anindilyakwa Land Council of Groote Eylandt, but we also rely on corporate and individual grants and donations to operate.

If I donate to the MJD Foundation, is my donation tax deductable?
Yes, the MJD Foundation is approved as a Deductable Gift Recipient with the Australian Tax Office.

If I want to learn more about MJD, where do I go, who do I contact?
As a starting point, you can contact the MJD Foundation by either:

- phoning us on 08 8948 0757
- emailing us on info@mjd.org.au
- writing to us at PO Box 1745, Nightcliff NT 0814
- Visit our website at www.mjd.org.au

We have many resources available to help, such as:

- Medical Protocols for standard treatments of main symptoms of MJD http://mjd.org.au/125-medical-protocols.html
- Help Sheets for people caring for those with MJD http://mjd.org.au/124-carer-information.html
- Educational DVDs http://mjd.org.au/19-resources.html
- Genetic Education booklets - A “Guide for all Australians with MJD”
- Online e-learning MJD module for health professionals http://www.rahc.com.au/elearning

HOW CAN I HELP?

If I want to contribute to support people with MJD, what can I do? Who do I contact?
There are many ways you can help people with MJD and the MJD Foundation in carrying out their work. Visit www.mjd.org.au and click on the “How to Help” tab.